2014

Who Are You Going to Call? Primary Care Patients' Disclosure Decisions Regarding Direct–to–Consumer Genetic Testing

Katherine Wasson
Loyola University Chicago

Sara Cherny
Central Dupage Hospital

Tonya Nashay Sanders
Morgan State University

Nancy S. Hogan
Loyola University Chicago, nhogan@luc.edu

Kathy J. Helzlsouer
Mercy Medical Center

RESEARCH ARTICLE

Who Are You Going to Call? Primary Care Patients' Disclosure Decisions Regarding Direct–to–Consumer Genetic Testing

Katherine Wasson PhD MPH[1], Sara Cherny MS CGC[2], Tonya Nashay Sanders PhD[3], Nancy S. Hogan PhD RN FAAN[1], Kathy J. Helzlsouer MD MHS[4]

1) Loyola University Chicago, 2) Central Dupage Hospital at Cadence Health, 3) Morgan State Univeristy, 4) Mercy Medical Center.
*Correspondence concerning this article should be addressed to Katherine Wasson, PhD, MPH, Assistant Professor, Neiswanger Institute for Bioethics, Health Sciences Division, Loyola University Chicago, 2160 S. 1st Avenue, Bldg. 120, Room 284, Maywood, IL 60153.
E–mail: kawasson@lumc.edu

Acknowledgements. The authors would like to thank Bryanna Cox MS CGC and the Loyola University Health System Primary Care Department for their assistance in the early phase of our research.

Conflicts of Interest. The authors have no conflicts of interest to declare.

Funding. This research study was funded by a grant from Loyola University Chicago.

Abstract. *Background:* Direct–to–consumer genetic testing (DTCGT) offers risk estimates for a variety of complex diseases and conditions, yet little is known about its impact on actual users, including their decisions about sharing the information gleaned from testing. Ethical considerations include the impact of unsolicited genetic information with variable validity and clinical utility on relatives, and the possible burden to the health care system if revealed to physicians. *Aims:* The qualitative study explored primary care patients' views, attitudes, and decision making considerations regarding DTCGT. This article focuses on the disclosure decisions participants made regarding participation, testing, and results of DTCGT, a topic which arose as a secondary aim of the study. *Methods:* Through four longitudinal interviews (pre–test, results, 3 and 12 months post–test) we examined twenty primary care patients' decisions, expressed intentions, and actions regarding disclosure to immediate and extended family, friends and coworkers, and physicians about participation in and results of DTCGT. Individual interviews were analyzed using qualitative content analysis and a summative approach to describe the global themes. *Results:* Most participants disclosed to some immediate family; less than half disclosed to extended family; approximately half talked to friends. Most participants stated they would or might disclose to physicians about DTCGT and a few did. Conceptual themes that emerged from the data analysis include ambivalence about disclosure, consistency between intention and actual disclosure behavior and decisions, and conditional information sharing. *Conclusions:* Participants' intentional and actual disclosure patterns offer insight into how they view DTCGT, weigh results, and the potential impact of DTCGT.

Key words. Decision Making, Direct–to–Consumer, Disclosure, Genetic Testing, Primary Care

Introduction

Since the completion of the Human Genome Project, the marvel of the human genome has piqued the interest of the general public. Private companies cater to this interest by offering direct–to–consumer genetic testing (DTCGT) based on genome–wide evaluation of subtle differences in the genetic

sequence. DTCGT, which returns a wide range of genetic risk results with varying levels of analytic validity and clinical utility, offers a different model for obtaining genetic information than traditional clinical genetic testing, which is usually done for individuals identified as at high risk for specific diseases (Wasson, Cook, & Helzlsouer, 2006; Wasson, 2009). DTCGT offers risk evaluation for health disorders, physical traits, drug response, and carrier testing for Mendelian disorders. Due to the availability of DTCGT in the public arena, it is important to assess its impact on individuals, communities and wider society, which helps to inform appropriate education, policymaking, and clinical practice.

One aspect of assessing the impact of DTCGT is disclosure decisions individuals make regarding testing and results. Disclosure, the focus of this article, has been addressed in the context of genetic testing for single–gene disorders, especially conditions with high emotional and medical impact such as hereditary breast and ovarian cancer, fragile X syndrome, and Huntington's disease (Forrest et al., 2003; Forrest et al., 2008; Gaff et al., 2007; Klitzman, Thorne, Williamson, Chung, & Marder, 2007; Peters et al., 2011; Ratnayake et al., 2011). These studies address the content and accuracy of disclosure, to whom participants choose to disclose results of genetic testing, their concerns about disclosure, and effective communication practices. Ethical concerns have been raised about the impact of DTCGT on individuals, families, and health care providers as previous research indicated potential consumers would expect their physician to help them understand results, thus potentially burdening the health care system if uptake of testing increases (McGuire, Diaz, Wang, & Hilsenbeck, 2009; McGuire & Burke, 2008). In addition, decisions made on the basis of inaccurate or unproven DTCGT information, including false positives or false negatives, have been raised regarding consumers and could also apply to their blood relatives if results were shared (Wasson, Cook, & Helzlsouer, 2006). Consumers could reveal unwanted or disturbing genetic information, e.g. regarding hereditary cancers or Alzheimer's Disease, to relatives without their "consent" or recognition of the potential consequences for the other. Yet, little is known about how actual users of DTCGT decide to whom and when to disclose this information, if at all. There are few published studies that address the details of disclosure choices regarding DTCGT participation and results in particular participants' decision-making choices and reasoning over time.

Limited data are emerging regarding DTCGT disclosure or intent to disclose to family members and/or physicians (Bloss, Schork, & Topol, 2011; Gollust et al., 2012; Kaphingst et al., 2012; McGowan, Fishman, & Lambrix, 2010; O'Daniel, Haga, & Willard, 2010; Ormond et al., 2011; Gordon et al., 2012). The data from actual users of DTCGT are limited and do not paint a consistent picture. McBride, Koehly, Sanderson, & Kaphingst, 2010, state the necessity of research "characterizing patterns of information sharing among the social network" and that "methodologies that provide an understanding of the family social structure and how that structure affects and is affected by genetic information are needed."

We conducted an exploratory, qualitative study of primary care patients' attitudes and views about DTCGT, aiming to learn more about what factors or considerations influence their decision–making processes regarding whether or not to participate in DTCGT. The importance of disclosure decisions arose during data analysis. The act of disclosure is characterized as how participants use and disseminate DTCGT information, e.g. to whom participants reported talking about the DTCGT research, testing or results. Our reasons for focusing on disclosure choices are three–fold. First, we aim to explore disclosure as a behavior or action that can be documented over time, monitoring intention and consistency in participants. Secondly, following disclosure patterns longitudinally offers insight into participant decision making, painting a more nuanced and detailed description of their processes. Disclosure directly reflects part of participants' motivation for pursuing DTCGT, and in some cases disclosure itself is a reason to pursue testing. Finally, we suggest that the act of disclosure is an identifiable indication of participant preferences. How participants use and disseminate genetic information

helps us understand what they find important—or on the other hand, insignificant—for themselves and/or their families. Understanding patient decision–making processes and preferences regarding DTCGT disclosure may inform those involved in genetics and primary care, and discussions in wider society about the impact of this testing.

Methods

Participants

This exploratory qualitative study was reviewed and approved by the Institutional Review Board at Loyola University Chicago. We targeted a primary care patient population for several reasons: (1) There was at the beginning of the study (August 2009), and remains, a lack of published research on primary care patient decision making about and reactions to DTCGT; (2) This patient population offered insight into the decision making of a potentially more diverse sample than early studies, which included social networkers and early adopters; (3) We hypothesized they might use their primary care physician as the gateway contact with the medical field, particularly when receiving DTCGT results; and (4) Recruited participants would already have an identified physician in case of medically relevant findings.

The study was conducted in two phases. Phase One included four focus groups of patients recruited from a primary care waiting room at an urban academic medical center. Patients were approached in the waiting room, informed about the study, and asked if they would consider participation. Those answering "yes" provided contact information and were later re–contacted and invited to participate in a focus group. Eligibility criteria included being 18 years old, English speaking, and not having had prior genetic testing (except for standard pre–natal genetic screening, which is widespread). A total of twenty–nine primary care patients participated in this phase and results are reported elsewhere (Wasson, Hogan, Sanders, & Helzlsouer, 2012). Here, we report results from Phase Two (December 2009 to June 2011), in which twenty participants provided informed consent for research and testing, underwent DTCGT, received results and participated in two additional follow–up interviews. All participants from Phase One were informed of Phase Two after each focus group and asked if they would consider participating. Sixteen participants (55%) from Phase One chose to undergo DTCGT in Phase Two; four additional participants were recruited directly from the primary care waiting room. Based on other qualitative studies, we determined that 20 participants would capture a range of views and attitudes needed to reach data saturation (Madsen, Holm, & Riis 2007; Schmidt, 2010). Because almost half of the individuals who participated in Phase One chose not to undergo DTCGT, data from each phase are reported separately (Wasson, Hogan, Sanders, & Helzlsouer 2012; Wasson, Sanders, Hogan, Cherny, & Helzlsouer, 2013).

Data Collection

Data were gathered through four individual interviews occurring over approximately 12 months during the: 1) Informed consent session, where a saliva sample was provided for DTCGT; 2) Receipt of results 4–6 weeks after the informed consent session; 3) Three months post–results, and; 4) 12 months post–results. Interviews one and two included a genetic counselor to answer questions and review results with the participant. In interview one, the genetic counselor fielded questions about the nature of testing and the nuances of the consent documents. In interview two, the genetic counselor pointed out any increased risks listed in the results, and discussed the genetic etiologies of these findings and others, allowing the participant to explore their results pages and ask any questions.

All 20 participants provided a saliva sample for analysis after their individual informed consent session, which included the study consent process and the 23andMe online consent form. The 23andMe test kit was used because it included a broad range of test types: carrier status for known single–gene conditions, common disease associations, drug susceptibility, and physical traits such as freckling

and earwax type. Further, these tests have widely varying clinical specificity and results range from clinically actionable (e.g. BRCA mutations) to potentially interesting but medically irrelevant (e.g. eye–color or baldness). Lastly, the 23andMe kit was the least expensive DTCGT available at the time. The cost of testing ($299–499) was covered by the study.

At each interview, participants were asked open–ended questions about reasons for participating in the testing, their decision to test, expectations, concerns about testing, and with whom they had or would discuss their participation and/or results. Interviews 3 and 4 (three and twelve months post–results) also included questions on participants' overall reaction to the testing process and whether testing had had any impact on them physically, emotionally/psychologically, or on their relationships/family, and lifestyle/behavior. They were also queried about their willingness to pay for and/or endorse this type of genetic testing. All twenty participants completed the first three face–to–face interviews and 17/20 completed the 12–month interview in person or by telephone. All interviews were audio recorded and transcribed verbatim. Participants received fifty dollars and a parking pass for each interview.

This analysis focuses on responses to specific questions asked to capture participants' intentions to disclose information about taking the DTCGT and actual disclosure decisions over time. Disclosure patterns and decisions arose as a key area during the data analysis as participants articulated and reflected on their decisions and actions over time.

Data Analysis

Qualitative content analysis was used to analyze responses to specific disclosure questions. Conventional content analysis is an inductive approach where the researchers immerse themselves in the data and allow categories to emerge from participants' own words, rather than imposing preconceived categories on the data. Researchers also used a summative approach to gain a sense of the global trends in disclosure in the data (Hsieh and Shannon 2005; Morgan 1993).

Two of the researchers (Sara Cherny and Katherine Wasson) read all the interview transcripts to familiarize themselves with the data and made notes about potential categories for analysis. They used this initial reading to become familiar with the context in which the disclosure questions were answered. Next, they again independently read the specific disclosure questions in detail and identified potential categories for recording to whom participants intended to and actually did disclose DTCGT participation and/or results at each interview. They then determined the final categories (e.g. intention to disclose participation/results, may disclose, did disclose, did not disclose, will not disclose). Both researchers met in person and together re–coded each participant's responses to the disclosure questions, resolving any differences in coding during this session. This process allowed the researchers to evaluate more clearly participants' stated intentions and actions about disclosure and emergent patterns over time.

In addition to specific categories of disclosure, i.e. to whom participants disclosed, overarching themes also emerged during the data analysis which shed light on why participants chose to (or not) discuss the testing and results with particular individuals or groups. Both researchers read the transcripts independently and met regularly to discuss the potential themes, resolve any discrepancies, and agreed the final themes for inclusion in the analysis. No qualitative software was used.

Results

Our sample was 50% African American, 50% Caucasian, and 60% female. Participants' age ranged from 29 to 63 (average of 49.5 years), and 65% were married. The proportion of African Americans in our sample is higher than in the general population (12.6% per US Census website), and participants had lower education levels as compared with participants in other currently published studies on the impact of DTCGT (McGowan, Fishman, & Lambrix 2010; Kaufman, Bollinger, Dvoskin, & Scott, 2012; Bloss et al., 2010; O'Daniel et al., 2010). All participants were high school graduates; 30% had college or post–graduate degrees. Fifty–five percent of participants carried private insurance, 40% had Medicare or Medicaid, and 5% were uninsured (See Table 1).

Table 1

Individual Testing	
N=20	
Age	
Range	29–63
Mean/Average	49.5
Income	
Less than 5,000	10.00%
5,000–9,999	0.0%
10,000–24,000	30.0%
25,000–49,999	25.0%
50,000–74,999	15.0%
75,000–above	20.0%
Sex	
Female	60.0%
Male	40.0%
Education	
High School Grad	30.0%
Some College	40.0%
College Grad	25.0%
Post Graduate	5.0%
Marital Status	
Never Married	15.0%
Married	65.0%
Separated/Divorced	10.0%
Widowed	10.0%
Race/Ethnicity	
Black	50.0%
White	50.0%
Insurance	
Private	55.0%
Medicare/Medicaid	40.0%
None/Uninsured	5.0%

The analysis of participants' disclosure of DTCGT information revealed four groups of people in their social network to whom they may have intended to disclose or actually disclosed participation and/or results of testing (See Figure 1). Circle 1 includes those in closest proximity to the participant and most likely to be affected by genetic health information, i.e. their immediate family (spouse, children, parents, siblings). Circle 2 includes extended family who might also be affected by genetic health information (aunt, uncles, cousins, grandparents, in–laws). Circle 3 includes friends, coworkers, and other non–family. Physicians were considered and analyzed separately as they are not blood relatives and may not be "close" to the participant. Due to the physician's role in maintaining the patient's health and wellbeing, participant choices regarding disclosure of DTCGT results to him or her may be different from their choices regarding their family and social network.

For the analysis of disclosure behaviors here, interviews 1–2, the informed consent and initial results session, are usually grouped together, and interviews 3–4, which were 3 and 12 months post–results are grouped together. Participants did not have the opportunity to report whether they revealed results to anyone until interview 3, though they could have mentioned participation in the study. Overall, by interviews 3–4, almost all participants (19) had talked to some immediate

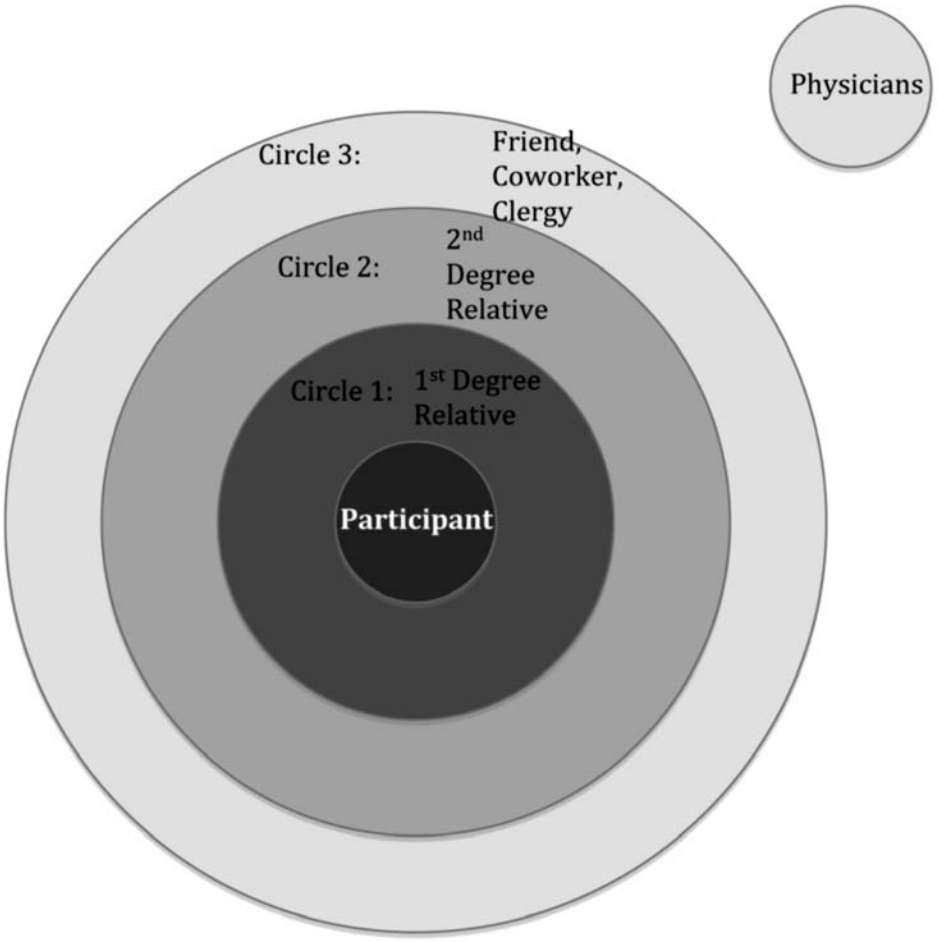

Figure 1 Family and social networks.

family (Circle 1); only one participant talked to no one. Some (six) only talked to those in Circle 1 and for a few, this meant only their spouse (3). In addition, some (seven) disclosed to extended family and approximately half of all participants (11) talked to people in Circle 3. A majority of participants disclosed to immediate family and friends, while a minority disclosed to extended family. Only a small number of participants (five) talked to people in all three circles.

Disclosure to Family

Disclosure to family included the first two circles in the analysis. Over half of participants reported being married, and all of these reported sharing both participation and results with their spouse. Most participants expressed an intention to disclose information about participation in DTCGT to others in Circle 1 in addition to a spouse during interviews 1–2, and all but one did disclose to immediate family members by interviews 3–4. For example, this participant noted in the first interview that disclosing to family was intended and would be important.

> Definitely, definitely the family. We are so close knit of a family. We talk about everything and in the group discussion with the people that I had a couple weeks ago, I was talking about how it would be good if you definitely talked with your family. A lot of people were kind of against that, a little shady on it, and I thought, well, if you share with your family something may come up and they will tell you, "Well hey, you know, your great, great aunt had this and you know you never knew about it," so when you talk with the family things do come up that would actually help you and your family and your kids and their kids. So, definitely will share it with my family. (Participant 12: Interview1)

Regarding Circle 2, some participants (seven) indicated by interview 4 that they had talked to different extended family members about their experience with DTCGT. Interestingly, only approximately half of this group had indicated a prior intention to disclose to extended family in interviews 1 and 2. Nearly twice as many participants reported talking to extended family after receiving results versus prior to testing. A few participants (three) demonstrated a lack of intention to disclose participation or results beyond their spouse and did not.

Disclosure to Friends and Coworkers

While only some (seven) participants noted an intention to disclose their participation to Circle 3 during interviews 1–2, a majority (12) had talked about participation or results with them by interviews 3–4. For example, this participant reported talking to multiple people about DTCGT.

> Oh, everybody that knows I'm coming here today. Monday at work, with my wife and a couple of my friends that I'll see over the weekend that I've told I was doing this. They all find it very interesting, they really do. (P6:I2)

Some (eight) chose not to disclose results to friends or co–workers, noting the information was personal and not necessary or appropriate to discuss with others.

> They weren't family and they weren't my doctor . . . I figured my results for something like that is personal. (P11:I4)

Disclosure to Physicians

During interviews 1–2, less than half (eight) of participants expressed an intention to discuss participation and/or results with their physician in response to an open–ended, general question about disclosure; a specific prompt for disclosure to physicians was included in interview 3. Of those expressing intent to talk to their physician, all but one did disclose by interview 4. An additional eight participants said they might disclose participation or results to their physician (interviews 1–3) and only one did. A few (four) participants expressed no intention to talk to their physician about participation in DTCGT and only one did disclose by interview 4.

Participant Disclosure Themes

The data analysis revealed three overarching themes that were significant in capturing participants'

intentions and actions regarding disclosure across the different groups. These included ambivalence or nonchalance about disclosure, consistency between intention and actual disclosure behavior and decisions, and conditional information sharing.

Ambivalence and Nonchalance about Disclosure

Several of our participants expressed ambivalence or nonchalance about disclosure of their results, especially to physicians. Ambivalence is reflected in the difference between those indicating they will (intention) or might discuss participation and/or results with physicians and actual disclosure to physicians by interview 4. Sixteen of the twenty stated by the third interview that they would or might tell their doctors about their participation or results at some point in the future. By the fourth interview (12 months post–test), only half (eight) of these had shared results with their physician; the majority of these had expressed a clear intention to do so earlier.

In the third and fourth interviews, some participants explained that they had not had an opportunity or reason to see their doctor, and one participant simply forgot. No participant reported making an appointment with a physician specifically to discuss participation in or results of DTCGT. No participant stated that their test results were concerning or significant enough to have an emotional or psychological effect on them. At the fourth interview, some participants did not remember what their results were.

> I haven't spoke to them at all . . . I don't go to the doctor's office that much. . . . Probably, I probably will. Not saying that I won't, you know, but it might come up in a conversation you know. (P10:I3)

> I have not. I haven't seen my primary care physician, and I don't know that I will. When I went to the website to see about this particular one I thought even if my kids do have it now, from what, which I don't think they do, but all they can do at this point seems to be the same kind of healthy living practices that we do, so I don't know that there's a need for that. I might mention it to my primary care physician just that I had the test or was in the research study or something. (P4:I3)

Ambivalence was more difficult to assess with regard to family and friend circles. There did not seem to be the same pattern of having intent to disclose and then not doing so. On the contrary, participants seemed to disclose to various members of their social circles without having previously demonstrated intent to do so.

Consistency

For a minority of participants, their intent and actions were clearly consistent regarding physicians: all participants who had shared results with their physicians by the third interview had stated their intent to do so in the first and/or second interviews. One participant clearly noted in the first interview that he would share the results with his physician. He did so by the third interview, although it seems he wanted more time spent on the results:

> (First interview) When I get the results of this I will bring the results over to my general practitioner regardless of what . . . (P16:I1)

> (Third interview) Well I briefly mentioned it to my doctor . . . I'm not sure if it was really discussed to the full extent. So he took a quick copy of it and it was put in the file. Of its worth for the information, I'm not sure if it was actually used to the full extent. If I had more time I could have, or we may have, I don't know. (P16:I3)

Three participants said they had no intent to discuss participation or results with their doctors, and none of them did disclose to their physicians.

Regarding intention and disclosure to immediate family, participants were overwhelmingly consistent. Most expressed the intention to disclose to someone in Circle 1 early on and then stated that they did during later interviews. Regarding extended family, the picture is more inconsistent. Only half of participants who reported disclosing to them in interviews 3–4 had indicated an intention to do so in interviews 1–2. When comparing

intention to actual disclosure to someone in Circle 3, friends and coworkers, the picture is also inconsistent. Again, only half of the participants who reported telling this group by interview 4 had stated an intention to talk to them in earlier interviews.

Conditional Information Sharing

For some participants, the motivation to share information was in some way conditional, in that their disclosure would depend on the nature, content, perceived impact, and/or perceived clinical utility of the DTCGT information. This conditionality applied both to decisions to disclose and not to disclose, and to disclosures to individuals within family, social circles and physicians.

Results content

For some participants, the content of results was a key factor in weighing their decisions to disclose or not. In the first interview, one participant stated that disclosure would depend on the result content as well as whom the results might affect:

> Well it all depends on what the results say. I mean, if it's something that is a definite that could possibly be something in the future as far as just affecting me, then probably not. But if it's a marker that can say, well it's genetic linked and it can be given down the line, then I probably will. You know if I start to see symptoms within the kids I probably will make mention, but right now just because everything is so fairly new, I probably wouldn't go into details right now. (P3: I1)

This same participant, after seeing her results, stated that the questionable accuracy and low impact of the results were a reason to not disclose:

> I probably won't, because it was nothing really to discuss . . . nothing showed up, so it probably wouldn't be worth mentioning. I mean, down the line if something like this comes available again and, you know, they say that it's a 100% proof that this is what this is, and this is because of this, then I may partake in something like that again just to have a definite answer to say yes, this is because of family history and this is what your risks may be. But because this is something preliminary, it's like not even worth mentioning at this point I don't think. (P3:I2)

Participants who had children sometimes commented on the benefits and limitations of sharing results with their children. For example, when asked with whom she might discuss results in the future, one participant said "my two children and probably my mom, it all depends" and then clarified:

> I have to hear what the results are. Most likely whatever it—I've been through worse—so whatever it is I believe I will discuss it with them. My daughter kind of takes things kind of hard, so that's why I said maybe. My son may be more so than her. I would have to see. But I'm believing in good results, good results from that. (P8:I1)

> Being a breast cancer survivor and having two teenage daughters I thought that any information that I could glean from the testing would be important, and that was primarily the reason why I did it. (P15:I3)

Influence of anticipated response

In weighing whether to disclose DTCGT information, some participants discussed how they expected the other person would perceive the DTCGT information, whether positively or negatively, or if they might find it useful.

> . . . I discussed it with my husband and he agreed, he says you have nothing to lose, you have nothing to lose, you have more to gain just, you know, just to find out what's what since these things do run in your family and how high of risk are you for these diseases or different types of things. So he suggested it. My daughters, I talked to them about it to. I let them know, you know, what my findings were and they said this is good, something they may want to do in the future. (P14: I3)

A few participants chose not to disclose based on an anticipated negative response from others.

> They don't even know that I'm even here doing the second one because my step daddy you know he's a big jokester, he jokes a lot, he said he wouldn't do it so I don't think I want to talk to him about it, because I don't want him to get my mom nervous or nothing like that, so I didn't

> talk to nobody about this one here so they don't even know I'm here. (P1:I1)

Others anticipated that the response of their family members to the results would be positive.

> My mother, interestingly enough, who is 95 years old said, "oh I'm so glad you're doing it." Well this is typical of her, but, "I'm so glad you're doing it, you'll have to tell me the results." (P4:I2)

> Of course my family knows. We have several chronic disorders in my family that I think are genetic or have a genetic component and I think everybody just agreed that "Hey, if you have a chance to do this, do it and find out what you can find out." (P4:I1)

Influence of Risk Perception

Another factor that affected disclosure choices was the participant's perception of the risk of developing the diseases addressed in the DTCGT results. Some participants had specific health or disease risk concerns, such as heart disease, cancer or diabetes, while others were more broadly concerned with receiving "bad" or high risk results. For a few, results showing little to no risk of disease, or "good results", decreased their likelihood of disclosure. After receiving her results, one participant said:

> Probably no one. I don't think that there was anything that was, that I feel that I need to go home and say, "Oh," you know. (P18:I2)

For others, low risk estimates in results increased their likelihood of disclosure because they could reassure family that they were not at high risk for developing diseases:

> When I go home I'll discuss it more with my husband and then, you know, talk to maybe my mom . . . she's not very optimistic. So this way I can tell her, "Look, I'm fine." (P2:I2)

> . . . I think I stated the last time, I mean since because I'm the guinea pig, so–to–speak of the family, it's like I didn't want to alarm them ahead of time and then have everybody on pins and needles waiting on results. I shared with my brother after the fact and after I got the results and everything was clear I brought it to their attention that there's something they need to be doing as far as for themselves and their kids and wife. Everybody needs to do it, period. I mean I really strongly think still it needs to start at birth and just have it in their file from birth. I mean I strongly believe that. I mean that way it will narrow down any type of problems if it's done at birth. (P13:I3)

Clinical Utility

Decisions about disclosure to physicians were sometimes related to a family history of disease, or if the testing identified specific areas of health concern. Some participants perceived that sharing the results with their physicians could help their care directly or prophylactically.

> Oh I will, definitely just for his opinion. . . . he's very thorough, and just to get his opinion and let him know that that information is there if I'm ever going to need it for whatever reason. But I will tell him. (P6:I2)

Only a few participants viewed their results as clinically useful. For example, a result showing increased sensitivity to the drug Warfarin increased the desire of a participant to share the results with her physicians:

> To help them be able to treat me better medically, and one of the tests showed sensitivity that my primary care quickly put on my medical profile because he's never seen it, but if it ever happens he's glad to know. (P7:I3)

Some participants gave specific reasons why they decided not to share the results with their doctor including the results not showing new or concerning health information.

> Because it said that I didn't have anything wrong so I didn't. (P2:I3)

> I think it was important, you know, but it wasn't anything I think that he already didn't know so I didn't bring it up to him yet. (P8:I3)

Participants intending to talk to their doctor generally expressed confidence in their physicians' ability to incorporate the results in to their care plan.

> There's all the little aspects of it, so if it's something that could be used, why not? It's just like other tests, it's like a CAT scan or an MRI, we would use it. That's all. (P16:I3)

The reality of talking with their physician was somewhat different. For example, one participant expressed confidence in her physician's ability to use the test results during her third interview. In the fourth interview, after she had discussed the results with her physician, she expressed disappointment in her doctor's reaction:

> After the test when I had like a physical, I had mentioned like I had these screenings, these tests, the study done. And it seemed like she just sort of brushed it off. (P14:I4)

Of the eight participants noting they had minor children, three participants, all female, felt that the results would be clinically relevant for their children. They all stated that they would speak or were considering speaking to their children's physician, independent of their choice about discussing the results with the children themselves. For example, one stated that she would discuss it with her children "when and if they're ready, but with their primary care, yes." (P7:I2)

Another was considering it:

> Stuff that comes back where you are more likely to get this and there is something that I have passed on, I don't want the children to know. Maybe the doctor, like maybe their pediatrician should know. (P9:I1)

Discussion

This study highlights disclosure decisions in primary care patients undergoing DTCGT. Participant decisions about disclosure seem to depend on multiple factors—the type of information anticipated and provided, the actual results, personal reaction to DTCGT information, and perceived reaction to results by others. Participants talked to a variety of people, including immediate and extended family members, as well as non–family members and physicians.

Some of the parties to whom participants disclosed would potentially be at risk for the same conditions identified as risk factors for the participant. One individual participating in DTCGT could lead to further interest in or actual testing of family members. If based on unreliable or inaccurate DTCGT results, there could be unnecessary anxiety for relatives or unnecessary cost to the patient and health system. In addition, the participant could reveal unwanted health information, e.g. about hereditary cancers or Alzheimer's Disease, to relatives without their full understanding of or desire to obtain that information.

For example, family cascade testing is common in genetic counseling practice for families in which an individual has been identified as having a specific disease risk or genetic condition. Targeted testing is systematically offered to subsequently more distant family members from the original tested individual. The resulting cascade could have positive or negative impact on those individuals and/or the health care system depending on the results and risk estimates given, personal and professional interpretation of results, and any subsequent actions taken by the tested individual(s) and/or their physician(s). The potential effect of DTCGT relates to the statement made by McBride and colleagues (2010) regarding the need for understanding of kinship networks. It will be important to identify more clearly these various familial and communal disclosure patterns to evaluate further the impact of DTCGT on individuals, their social networks, physicians, the health care system, and society more broadly.

Ambivalence/Nonchalance

In our study, ambivalence regarding disclosure was most evident with regard to disclosure to physicians. Other studies have addressed physician disclosure within their populations. Gollust and colleagues (2012) and Bernhardt and colleagues (2009) found high levels of intent in their respective populations: 92% and 96% of potential DTC genetic testers predicted they would talk to their doctors about their results. In comparison, less than half (8/20) of our sample expressed specific intent to disclose participation in and/or results of DTCGT with their physicians at any point in the study. Only five of our participants clearly indicated intent to disclose to their physicians in the first interview,

prior to testing (and did so). Additionally, less than half (8/20) said they might disclose to their physician, and of those, 6 had not by the time of Interview 4. Thus, we are cautious when comparing our participants, having used DTCGT as part of the study, to potential users of DTCGT who have not (yet) proceeded with testing, as there seems to be a gap between intent to disclose and actual disclosure decisions after testing.

With regard to actual disclosure of results, reported findings are varied. Our findings are most comparable to that of the Coriell Personalized Medicine Collaborative (CPMC) participants, where 25/60 spoke to a physician by the time of their interview and 14 more indicated intent to disclose in future (Gordon et al., 2012). Kaufman and colleagues (2012) reported that 28% of their population of DTCGT users talked to at least one healthcare professional, and 20% shared results with their primary care physician. O'Daniel and colleagues (2010) and Kaphingst and colleagues (2012) offer more dramatic examples, with 0% and 1% of their respective samples having spoken with a physician three months after participating in DTCGT or genetic susceptibility testing. It is important to note that there seems to be a significant difference between the intentions of potential users and the intentions and disclosures of actual users of DTCGT both in our study as well as in others. Our longitudinal format and qualitative analysis, allowing for investigation of both intended and actual disclosure decisions and choices pre–and post–test, may demonstrate a more nuanced participant thought process with regard to physician disclosure than previous studies.

If this general ambivalence is applicable to a broader population, it could inform the discussions about the potential burden on physicians from this type of testing and the necessity of medical professional involvement in DTCGT (Howard & Borry, 2012; McGuire & Burke, 2008). If results do not have a significant impact such that participants are not reporting them to their physicians or seeking help interpreting them, then there may not be a marked increase in physician time demands or additional testing and burden on the health care system as a result of DTCGT being available to the general public (Giovanni, 2010). It is important to reiterate that our participants received genetic counseling as part of their testing process, as the investigators viewed it as a necessary and ethical part of practice, which may have affected their desire to seek advice or recommendations from a physician, or simply to share their participation and/or results. If their reaction is because the genetic counseling played a significant role, then the findings identify a need for that process. Therefore, while there may not be an increased burden on physicians from DTCGT, there would be on genetic counseling services, which would not necessarily be included in DTCGT. Alternatively, participants may have received closure on or answers to their genetic susceptibility questions from the testing and, therefore, did not feel the need to discuss it further with physicians or other health professionals.

Conditional Information Sharing

Clinical Utility

For those participants who expressed ambivalence regarding their potential information sharing about DTCGT, we speculate that it may be related to their perception of clinical utility. Few participants reported finding their results clinically useful; some stated that the results were consistent with what they already knew about themselves or their families, e.g. increased risk for heart disease, or diabetes. If there was nothing new or interesting to learn from the results, participants may have been less likely to disclose them. Also, we think that participants might have different privacy expectations in relationships with family versus non–family. Disclosure choices may relate to varying levels of trust participants had in others with regard to how the discloser might want the information used. Participants who did not tell their physicians may not have trusted them with this type of information because of their

patient–physician relationship, previous experience in the health care system, or fear of genetic information in their medical record having implications for insurance coverage (Wasson, Hogan, Sanders, & Helzlsouer, 2012).

Consistency

The consistency between intention and actual disclosures to immediate family was notable. This information sharing is understandable because participants have regular contact with immediate family, providing the opportunity to talk about their experience with DTCGT, and they may also have thought that the testing might impact these family members more directly. In contrast, the patterns of intention and disclosure with extended family are inconsistent. Only half of participants who reported disclosing to them in interviews 3–4 had indicated an intention to do so in interviews 1–2. This situation may be because participants have less contact with extended family or are not as close to them as immediate family. It may also be related to the participant's intent with regard to participation in the study itself—if the participant was not seeking health information for his/her family, then there may not have been a strong impetus to reach out specifically to extended family members. It is worth considering further the implications of this disclosure pattern. Extended family was not necessarily alerted to the fact that the participant was undergoing DTCGT. If the participant had been given results which could or did affect family, extended family did not have the opportunity to consider the implications of or express their wishes regarding being told or not told the results. They were simply told about testing and/or results after the fact. For example, a cousin may not have wanted to know breast cancer results, but been unable to decline to know because the participant decided to share information post–results and without consultation before the disclosure.

Regarding Circle 3, friends and coworkers, there were twice as many participants who reported telling this circle by interview 4 than had expressed an intention to do so earlier. This finding might suggest that participants were unsure or ambivalent about what the testing would uncover. Once they received results, many saw them as not concerning and may have then felt more able to discuss the testing or results with friends. The results were perhaps viewed as non–threatening; instead they may have been seen as insignificant, recreational, or a novelty. We found it interesting that the breadth of disclosure to participants' social and family networks increased from their initial intention and that more talked to friends than extended family. We considered that participants might see friends more often than extended family as a reason that a majority of participants were willing to discuss this novel type of testing with friends over time. These patterns of disclosure may also suggest that views on risk of a privacy breach of genetic information are varied. Some participants were nonchalant about disclosure and others were clear they would not disclose to different circles. Further research is needed in larger populations to investigate these varied views and examine why and to whom participants of DTCGT choose to disclose.

Regarding physicians, participants who noted their intention to disclose to their doctor early on (interviews 1–2) did disclose, except for one person. This group was clear they wanted to tell their physicians about the testing and results. They were keen to get feedback from their physician, explore potential benefits of the DTCGT information now or in future, and trusted their physician to understand the results. They may have expected the physician to be more receptive or helpful, as some expressed disappointment in the reaction of their physician to

1 The version of direct–to–consumer genetic testing offered by 23andMe and used in this study is not currently available. The United States Food and Drug Administration is reviewing the tests and their reliability and accuracy and has requested the company cease to offer them to the public during that process. 23andMe continues to offer genetic ancestry testing direct–to–consumers.

their disclosure or lack of attention paid to them. Similarly, most of those participants who were clear in early interviews that they would not tell their physicians did not. They were clear that they did not see a benefit or wish to have the information enter the medical realm.

In contrast, most of the participants who were unsure or ambivalent about talking to their physicians had not disclosed to them by interview four. This choice may be due to the fact that they were not worried about results, did not view results as "bad", knew the tests were new and had variable accuracy and reliability, or had had their questions answered by the researchers or genetic counselor. Any or all of these factors may have influenced their decision not to disclose DTCGT or results to their doctors.

We hypothesize that there is more insight to be gained on the topic of consistency in intent and actual disclosures about DTCGT in a larger population, which could lead to further categorization and predictive ability.

Limitations of this Study

In evaluating our findings, it is important to note that this is a self–selected group of participants, and results are self–reported by them. Primary care patients who were willing to participate in this research may not be representative of other primary care patients. The cost of testing was covered by the study and the availability of free testing may have had an influence on participants' decisions to participate. The cost of testing with 23andMe has been as low as $99 since the study was completed, which may make cost less of a factor in uptake.[1] In addition, genetic counseling was provided to participants, which may have affected their disclosure decisions, particularly regarding physicians. Sixteen of the participants recruited for Phase 2 had participated in a Phase 1 focus group during which they had the opportunity to participate in a discussion of the issues surrounding DTCGT, which could introduce a bias.

When reporting retrospectively, participants were not always specific about the content of their disclosures, i.e. whether they disclosed participation and/or results to others. We reported specific data when we had consistent detail from a particular group of participants or category of people to whom they disclosed.

A qualitative, longitudinal perspective allows increased insight in to the process of decision–making and disclosure; however, it will likely be necessary to assess these actions over an even longer time frame and on a larger sample to assess more fully the impact of DTCGT and results on disclosure decisions and actions. Our findings are preliminary; further research is necessary to apply this information to a wider population.

Conclusions

In this study we explored and noted that disclosure intentions, decisions, and actions regarding DTCGT are carefully considered by participants, and they are willing to share the nuances of selective disclosure with researchers. It is possible to identify characteristics and considerations amongst our participants—consistency, ambivalence, reactions of others, content of results—that impact disclosure decisions, and might apply to or provide clues about a broader primary care population. Disclosure patterns may help assess the likelihood of uptake of DTCGT by members of a participant's social and family networks. They may also offer insight regarding the ethical issues for relatives who may receive unsolicited genetic information from participants and the potential burden upon physicians, genetic counselors, and the healthcare system of such information. In analyzing participants' disclosure patterns, this study offers insight into how they view the privacy of their DTCGT information. It will be necessary to evaluate further primary care patient intentions and actions to assess the potential long–term impact of DTC genetic testing on individuals, their families, communities, the health care system, and wider society.

References

Bernhardt, B., Gordon, E., Gollust, S., Keddem, S., Zayac, C, & Pyeritz, R. (2009). Why are people interested in personalized genomic risk information? *American Society for Human Genetics Annual Meeting Abstracts*.

Bloss, C.S., Schork, N.J., & Topol, E.J. (2011). Effect of direct–to–consumer genome wide profiling to assess disease risk. *New England Journal of Medicine*, 364, 524–34.

Bloss, C.S., Ornowski, L., Silver, El., Cargill, M., Vanier, V., Schork, N., & Topol, E.J. (2010). Consumer perceptions of direct–to–consumer personalized genomic risk assessments. *Genetics in Medicine*, 12, 556–66.

Forrest, K., Simpson, S.A., Wilson, B.J., van Teijlingen, E.R., McKee, L., Haites, N., & Matthews, E. (2003). To tell or not to tell: barriers and facilitators in family communication about genetic risk. *Clinical Genetics*. 64, 317–26.

Forrest, L.E., Curnow, L., Delatycki, M.B., Skene, L., Aitken, M., & Eur, J. (2008). Health first, genetics second: Exploring families' experiences of communicating genetic information. *European Journal of Human Genetics*. 11, 1329–35.

Gaff, C.L., Clarke A.J., Atkinson, P., Sivell, S., Elwyn, G., Iredale, R., . . . Edwards, A., (2007). Process and outcome in communication of genetic information within families: A systematic review. *Institute of Medical Genetics,* 15, 999–1011.

Giovanni, M.A., Lehmann, L.S., Green, R.C., Meckley, L.M., Veenstra, D., Murray, M.F. (2010). Health–care referrals from direct–to–consumer genetic testing. *Genetic Testing and Molecular Biomarkers*, 14, 817–819.

Gordon, E.S., Griffin, G., Wawak, L., Pang, H., Gollust, S.E., & Bernhardt, B.A., (2012). It's not like judgment day: Public understanding of and reactions to personalized genomic risk information. *Journal of Genetic Counseling*, 21, 423–32.

Gollust, S.E., Gordon, E.S., Zayac, C., Griffin, G., Christman, M.F., Pyeritz, R.E., . . . Bernhardt, B.A., (2012). Motivations and perceptions of early adopters of personalized genomics: Perspectives from research participants. *Public Health Genomics*. 15, 22–30.

Howard, H.C., & Borry, P.,(2012). Is there a doctor in the house? The presence of physicians in the direct–to–consumer genetic testing context. *Journal of Community Genetics*. 3, 105–112.

Hsieh, H.G., & Shannon, S.E., (2005). Three approaches to qualitative content analysis. *Qualitative Health Research*. 15, 1277–88.

Kaphingst, K.A., McBride, C.M., Wade, C., Alford, S.H., Reid, R., Larson, E., . . . Brody, L.C., (2012). Patients' understanding of and responses to multiplex genetic susceptibility test results. *Genetics in Medicine*. 14, 681–87.

Kaufman, D.J., Bollinger, J.M., Dvoskin, R.L., & Scott, J.A., (2012). Risky business: Risk perception and the use of medical services among customers of direct–to–consumer personal genetic testing. *Journal of Genetic Counseling*. 21, 413–22.

Klitzman, R., Thorne, D., Williamson, J., Chung, W., & Marder, K., (2007). Disclosures of Huntington disease risk within families: Patterns of decision–making and implications. *American Journal of Medical Genetics Part A*. 15, 143A, 16, 1835–49.

Madsen, S.M., Holm, S., & Riis, P., (2007). Attitudes towards clinical research among cancer trial participants and non–participants: An interview study using a grounded theory approach. *Journal of Medical Ethics.* 33, 234–240.

McBride, C.M., Koehly, L.M., Sanderson, S.C., & Kaphingst, K.A., (2010). The behavioral response to personalized genetic information: Will genetic risk profiles motivate individuals and families to choose more healthful behaviors? *Annual Review Public Health*. 31, 89–103.

McGowan, M.L., Fishman, J.R., & Lambrix, M.A., (2010). Personal genomics and individual identities: Motivations and moral imperatives of early users. *New Genetics and Society*. 29, 261–290.

McGuire, A.L., & Burker, W., (2008). An unwelcome side effect of direct–to–consumer personal genome testing: Raiding the medical commons. *The Journal of the American Medical Association.* 300, 2669–2671.

McGuire, A.L., Diaz, C.M., Wang, T., & Hilsenbeck, S.G., (2009). Social networkers' attitudes toward direct–to–consumer personal genome testing. *The American Journal of Bioethics*. 9, 3–10.

Morgan, D.L., (1993). Qualitative content analysis: A guide to paths not taken. *Qualitative Health Research.* 3, 112–21.

O'Daniel, J.M., Haga, S.B., & Willard, H.F., (2010). Considerations for the impact of personal genome information: A study of genomic profiling among genetics and genomics professionals. *Journal of Genetic Counseling*. 19, 387–401.

Ormond, K.E., Hudgins, L., Ladd, J.M., Magnus, D.M., Greely, H.T., & Cho, M.K., (2011). Medical and graduate students' attitudes toward personal genomics. *Genetics Medicine*. 13, 400–8.

Peters, J.A., Kenen, R., Hoskins, L.M., Koehly, L.M., Graubard, B., Loud, J.T., & Greene, M.H., (2011). Unpacking the blockers: Understanding perceptions and social constraints of health communication in hereditary breast ovarian cancer (HBOC) susceptibility gamilies. *Journal of Genetic Counseling*. 20, 450–64.

Ratnayake, P., Wakefield, C.E., Meiser, B., Suthers, G., Price, M.A., Duffy, J., . . . Tucker, K., (2011). An exploration of the communication preferences regarding genetic testing in individuals from families with identified breast/ovarian cancer mutations. *Familial Cancer*. 10, 97–105.

Schmidt, L.A., (2010). Making sure: Registered nurses watching over their patients. *Nursing Research*. 59, 400–406.

Wasson, K., Cook, E.D., & Helzlsouer, K., (2006). Direct–to–consumer online genetic testing and the four principles: An analysis of the ethical issues. *Ethics and Medicine*. 22, 83–91.

Wasson, K., (2009). Direct–to–consumer genomics and research ethics: Should a more robust informed consent process be included? *The American Journal of Bioethics*. 9, 56–8.

Wasson, K., Hogan, N.S., Sanders, T.N., & Helzlsouer, K.J., (2012). Primary care patients' attitudes and decision–making process regarding direct–to–consumer personalized genome testing. *American Journal of Bioethics Primary Research*. 3, 24–35.

Wasson, K., Sanders, T.N., Hogan, N.S., Cherny, S., & Helzlsouer, K.J., (2013). Primary care patients' views and decisions about, experience of and reactions to direct–to–consumer genetic testing: A longitudinal study. *Journal of Community Genetics*. 4, 495–505.